# CHAPTER III.8. COST OF DOWN SYNDROME

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# CHAPTER III.8. COST OF DOWN SYNDROME

## III.8.A Background

This chapter contains a discussion of the methods used and the results of estimating the direct medical costs incurred by individuals with Down Syndrome and the results of the analysis.[1] It does not include information on elements such as indirect medical costs, pain and suffering, lost time of unpaid caregivers, etc. The reader is referred to Chapter I.1 for a discussion of the cost estimation methods and cost elements that are relevant to all benefits estimates. In addition, Chapter III.1 contains information regarding the special characteristics of developmental defects, and a list of chemicals that may cause developmental abnormalities.

The costs presented in this chapter were current in the year the chapter was written. They can be updated using inflation factors accessible by clicking on the sidebar at left.

*Link to Chapters I.1 and III.1*
*Link to inflation factors*

### III.8.A.1 Description

Down syndrome occurs as a result of having three, rather than two, copies of chromosome 21 (hence the name "trisomy 21"). Mental retardation and a group of physical characteristics are commonly associated with Down syndrome. In addition, a number of serious defects in critical organs (e.g., heart, digestive system) are also commonly found in people with Down syndrome. The syndrome involves clusters of external physical anomalies, learning disabilities, and organ system anomalies. Physical anomalies and their prevalence (given in parenthesis) among Down syndrome children are: unusually small head (50 percent), excess skin folds on eyelids (50-70 percent), speckled irises in eyes (30-80 percent), narrow and short palate (60-90 percent), protruding tongue (40-60 percent), broad hands (70 percent), and other relatively minor changes in physical appearance. Down syndrome children usually have a flattened nose bridge, additional skin on the back of the neck, small or anomalous ears, abnormally formed fingers, and "simian" hand creases (Pueschel and Rynders, 1982).

The severity of mental retardation associated with this disorder varies considerably. Between 3 and 12 percent of Down syndrome children are profoundly retarded. Many of these individuals are unable to walk, talk, or eat without assistance. Approximately 25 percent are severely retarded, 55

[1] "Costs" in this chapter refer to direct incremental per capita medical costs, unless otherwise noted.

percent are moderately retarded, and 14 percent are mildly retarded. Seizures occur in five to nine percent of children, increasing with age. In adulthood, memory loss and reduced cognitive abilities are usually evident after age 45 in people with Down syndrome. Pathology studies have found structural changes in the brain similar to those found in Alzheimers disease patients (Waitzman et al., 1996).

Children with Down syndrome often have very early onset Alzheimer-type changes in cognitive ability. These changes may affect their functional abilities, and are observed when comparing the abilities of Down syndrome children with other children over the range of school ages. By young adulthood, the IQ scores of Down syndrome children have progressed from the range of low-mild to high-moderate retardation seen in early school years to a range that includes severe retardation (Oski, 1994).

Down syndrome is the most common chromosomal disorder observed in the newborn period and accounts for approximately one third of all chromosomal abnormalities. Overall, chromosomal abnormalities occur in approximately 1 in 200 live births and account for a sizable fraction of the malformations and neonatal deaths that occur. The frequency of Down syndrome is estimated to range from one in 700 to one in 1,000 (Oski, 1994). With the availability of prenatal testing, not all Down syndrome fetuses are brought to term. Although the total medical costs of Down syndrome and its incidence (which is measured in terms of live births) are thereby reduced, the individual and societal costs may be very substantial.

### III.8.A.2 Concurrent Effects

Associated organ system anomalies include congenital heart disease, duodenal atresia or stenosis, cleft lip or palate, fused digits, tracheoesophageal fistula, imperforate anus, failure of the testicles to descend, and incomplete development of neck vertebrae. Of these, the most common and serious are heart defects, which occur in approximately 29 percent of children with Down syndrome. Approximately 50 percent of adolescents and adults have mitral valve prolapse in the absence of symptoms of heart disease. The children usually also have poor muscle tone, increased joint flexibility, instability between the first two neck vertebrae, and an abnormally developed pelvis. Their musculoskeletal abnormalities often lead to degenerative changes in the joints. Thyroid function is often decreased (Waitzman et al., 1996).

Hearing loss affects approximately 60 percent of Down syndrome children, and 15 to 26 percent have moderate to profound hearing loss. Cataracts affect approximately 12 to 20 percent of children; strabismus affects 20 to 45 percent. Severe nearsightedness and deformity of the cornea are also common (Waitzman et al., 1996).

Down syndrome children have an increased risk of skin and lung infections, acute leukemia, and an impaired immune system function (Waitzman et al., 1996), and increased rates of testicular cancer and retinoblastoma (a childhood cancer) (Oski, 1994).

### III.8.A.3 Causality

Down syndrome occurs when the egg or sperm receives an extra copy of chromosome 21. This condition is hereditary in some cases and is also associated with maternal age. Chemicals that cause chromosomal abnormalities, especially interference with normal chromosomal replication and disjunction, may cause this type of genetic abnormality (Waitzman et al., 1996). Table III.1-2 in Chapter III.1 lists chemicals associated with genotoxic effects. A review of material in the references listed in Chapter III.1 will provide information regarding those chemicals that have been shown to cause non-disjunction, and other genotoxic effects that may specifically cause an abnormal number of chromosomes.

*Link to Chapter III.1, Table III.1-2*

### III.8.A.4 Treatment and Services

A variety of medical treatments and special services are required for Down syndrome patients. These vary widely, depending on the specific cluster of abnormalities occurring in an individual. The numerous structural abnormalities may require immediate attention in the postnatal period, or be corrected later in childhood if they are not immediately life-threatening. Some structural problems related to bone structure, muscles, and joints may require specialized equipment and physical therapy. Hearing problems and recurrent ear infections are often treated with pressure-equalization tubes and antibiotics. Due to the retardation associated with this syndrome, special education services are usually required. Many Down syndrome patients require lifelong services in the form of specialized housing and medical care.

### III.8.A.5 Prognosis

Down syndrome involves a cluster of effects. While some effects, such as facial appearance, do not change over time, others become more severe over time. For example, a deterioration in memory and other cognitive functions is seen in most Down syndrome patients over the age of 45 (Waitzman et al., 1996). Laxity in the ligaments often leads to degenerative changes in the joints, especially in the knees and spine (Semine et al., 1978; Mendez et al., 1988). The prevalence of seizures, which occur in approximately five to nine percent of Down syndrome patients (Waitzman et al, 1996), increases with age (McVicker et al., 1994). In addition to

increased morbidity over time, the lifespan of Down syndrome patients is likely to be shortened, due to the multitude of serious effects associated with this disease.

## III.8.B Costs of Treatment and Services

### III.8.B.1 Methodology

Waitzman et al. (1996) provide an estimate of the direct medical and non-medical costs of treating Down syndrome. They used the same methodology to estimate the costs incurred by individuals with Down syndrome as for all the birth defects for which they estimated costs. The methodology and relevant considerations are detailed in Chapter III.3, including discussions of direct and indirect costs, prevalence versus incidence, incremental costs, and concurrent effects. The analytic method, the sources of data and the limitations of the Waitzman method are also discussed in Chapter III.3. The methodology is outlined briefly here.

*Link to Chapter III.3*

To estimate the lifetime medical costs incurred by an individual with a birth defect, Waitzman et al. estimated the average lifetime medical costs for an individual with the birth defect. From this value, the authors subtracted the average lifetime medical costs for an individual without the birth defect. Because they estimated lifetime costs, they used an incidence-based approach. Ideally, they would have tracked the costs of the cohort members over time, until the death of the last cohort member. Because the members of the cohort were born in 1988, however, this tracking was not possible. Instead, estimates of the costs incurred at each age were based on estimates of per capita costs in the prevalent population of that age (see Chapter III.3, Section III.3.B.1.2).

*Link to Chapter III.3, Section III.3.B.1.2*

This method has two important implications. First, Waitzman et al. estimated the costs incurred by individuals with birth defects, including all medical costs incurred, rather than the cost of the birth defect per se. These cost estimates therefore include the costs of concurrent effects (unlike the costs reported for many of the diseases in this handbook). This method yields a more comprehensive assessment of total costs than would be obtained if only individual effects were evaluated. This method is of particular use in valuing the avoidance of birth defects because they very frequently occur in clusters within an individual. As Waitzman et al. note, however, the costs of associated anomalies are included as part of the estimate of the costs incurred by an individual with a given birth defect. These cost estimates therefore cannot be aggregated across birth defects because of the possibility of double counting.

Second, the Waitzman et al. method estimates the *incremental* costs for individuals with birth defects — that is, the costs above and beyond the average costs that would be incurred by individuals without the birth defect.

Waitzman et al. (1996) estimated three categories of costs incurred by individuals with Down syndrome: direct medical costs, direct nonmedical costs, and indirect costs.[2] Direct medical costs, specifically inpatient care, outpatient care, pharmaceuticals, laboratory tests, X-rays, appliances, and long-term care are included in the cost estimates shown in this and other chapters (Chapters III.3 through III.8) based on the work of Waitzman et al. Nonmedical direct costs, specifically developmental services, and special education are also included in this handbook.

The Waitzman estimates of the costs incurred by individuals with Down syndrome are based on the costs of this birth defect in California across many ages, and its occurrence in a large cohort of children born in California in 1988. California's ongoing birth defects monitoring program provides an excellent source of data. The California data sets were linked with other national data sets so that Waitzman et al. could estimate the incremental costs associated with Down syndrome.

The method of calculating the expected lifetime incremental costs for an individual with a birth defect — i.e., the average lifetime cost per case — is the same for all the birth defects considered by Waitzman et al. The expected per capita cost at age *i*, $PCC_i$, for an individual born with the birth defect is the probability of surviving to age *i* (among those individuals born with the birth defect), $ps_i$, times the per capita cost among individuals who do survive to age *i* ($PCPREV_i$, measured in the prevalent population):

$$PCC_i = (ps_i) \times (PCPREV_i) .$$

Waitzman et al. estimate per capita costs in the prevalent population of age *i*, $PCPREV_i$, in two different ways, depending on data availability (see Chapter III.3).

*Link to Chapter III.3*

[2] Indirect costs are not generally discussed in this handbook and so are not included in this chapter. The reader may wish to consult Waitzman et al. (1996) for information on these costs.

The present discounted value of expected per capita lifetime costs of the birth defect, PCCOBD, is just the sum of these expected age-specific per capita costs, appropriately discounted (as explained more fully in Chapter III.3):

$$PCCOBD = \sum_i PCC_i/(1+r)^i .$$

### III.8.B.2 Results

Waitzman et al. (1996) estimate the total lifetime medical costs of Down syndrome as outlined in the following tables, updated from 1988 to 1996 dollars based on the medical care cost component of the Consumer Price Index (1996:1988=1.6465). Table III.8-1 shows the annual per capita medical costs associated with Down syndrome by age group.

**Table III.8-1: Annual Per-Capita Medical Costs of Down syndrome by Age Group (1996$)**

| Condition | Age 0-1 | Age 2-4 | Age 5-17 | Age 18+ |
|---|---|---|---|---|
| Down syndrome | $27,265 | $5,577 | $2,231 | $7,529 |

The medical cost of the average population was then subtracted from these costs to obtain incremental costs. Waitzman et al. (1996) discounted these costs using three different discount rates: two percent, five percent, and ten percent. Although these discount rates do not match the standard EPA rates used in many other chapters in this handbook (zero percent, three percent, five percent, and seven percent), there is insufficient information provided in Waitzman et al. (1996) to allow a conversion to discounted costs using standard EPA discount rates. This problem exists in all chapters based on the Waitzman et al. data (i.e., Chapters III.3 through III.8).

The present discounted values of average per capita lifetime incremental costs, using discount rates of two percent, five percent, and seven percent, are listed in Table III.8-2 below. Direct medical costs and direct non-medical costs, including developmental services costs and special education costs, are listed separately. The sum of per-capita direct medical and nonmedical costs provides an estimate of the total per-capita costs incurred by individuals with Down syndrome.

**Table III.8-2: Per-Capita Net Medical Costs, Nonmedical Costs, and Total Costs of Down syndrome (1996$)**

| Condition | 2% | 5% | 10% |
|---|---|---|---|
| Net direct medical costs | $141,596 | $90,556 | $64,212 |
| Net direct nonmedical costs | | | |
| Developmental services | $67,645 | $35,439 | $19,089 |
| Special education | $144,138 | $109,622 | $72,854 |
| Total Costs | $353,379 | $235,617 | $156,155 |

The costs presented in this chapter were current in the year the chapter was written. They can be updated using inflation factors accessible by clicking below.

*Link to inflation factors*